



Rhode Island Commission on Women

Position Paper on the Ethical, Legal and Social Implications (ELSI) of Genetic Testing

This position paper focuses on the ethical, legal and social implications (ELSI) of genetic testing, which have arisen from advances made by the U.S. Human Genome Project (HGP). A previous Rhode Island Commission on Women (RICW) position paper covered health issues related to genetics.

It is the position of the RICW that genetic testing must be accessible, of high quality, and voluntary. Educating women and providers about genetic testing will provide knowledge to make crucial decisions concerning their health. Informed consent must be given prior to testing, and strict patient confidentiality must be maintained. Medical follow-up and counseling are recommended before and after testing if a genetic mutation is found. Ethical, legal and social implications inherent in genetic testing must be adequately addressed to prevent abuse.^{5, 6, 7} Currently, there are no stated consequences for violations of the Rhode Island law related to genetic testing in the areas of informed consent, confidentiality, genetic discrimination in employment, and health insurance.^{8, 9} Standards and guidelines must be established to address violations.

The National Human Genome Research Institute (NHGRI) has developed positions on informed consent in genetics research, genetic privacy, genetic discrimination, and genetic determinism.^{10, 11} Genetic determinism is when an individual or society identifies a person by their genetic makeup. Using that genetic information to determine the relative worth of an individual can lead to discrimination. Genetic discrimination is when an individual's access to normal rights is curtailed due to the possible risks of a genetic malady. Many legal and ethical issues are raised by this misuse of genetic testing, including employment discrimination, loss of health care coverage, discrimination in reproductive choice, and even unnecessary surgical interventions.

An ELSI working group was formed by the NHGRI in 1998 to stimulate public discussion and overall awareness of the issues raised by the mapping and sequencing of the human genome and to develop public policy.¹² The NHGRI devised four important policy options to guide Americans and their policy makers in making informed decisions about ELSI issues^{13, 14}.

1) Avoiding misuse of genetic information. Proper use and interpretation of genetic testing is necessary to avoid the incorrect understanding and possible misuse of the information. A partnership between the National Action Plan on Breast Cancer and the ELSI Working Group made policy recommendations for preventing genetic discrimination in health insurance, the work place, the Federal legislature, and the Health Insurance Portability and Accountability Act of 1996. In April 2003, HIPPA extended protection for confidentiality of medical records. These policies have been enacted to protect people against genetic discrimination in health insurance, prohibiting insurers from using genetic information as a pre-existing condition and from denying or limiting coverage in-group markets.¹⁵ As noted above, while Rhode Island has a genetic discrimination law, there is no enforcement if infractions of this law occur.

2) Responsible clinical use of new genetic technology. Principles for the responsible clinical use of genetic testing have been developed.¹⁶ The impact of genetic testing and genetic counseling on individuals, families and society can be profound. Concerns have been raised about the quality control of genetic testing, in terms of its safety, effectiveness, accuracy and correct interpretation. Genetic testing should be done only on a volunteer basis, and caution is necessary in the use and interpretation of genetic testing for individuals with pre-symptomatic genetic disease.

3) Ethics of genetic research. When patients participate in genetic research studies, ethical issues arise related to informed consent, as well as the design, conduct, participation in and reporting of the research. Institutional Review Boards have been established to protect patients participating in research. They can provide patients with a guide developed by the National Institute of Health's Office for Protection from Research Risks that discusses the patient's

right to protection of privacy and autonomy, and the welfare of a volunteer's participation in genetic research.

4) Education and training. Educating health professionals, policymakers and the general public on ELSI issues in genetic testing is important.¹⁷ NHGRI, in partnership with public and private agencies, supports efforts to create model education programs to educate students, consumers, healthcare professionals, appellate judges, and journalists about genetic technology.

The Advisory Committee on Research on Women's Health and the World Health Organization agree that women have the right to have access to pre-symptomatic genetic testing for breast cancer and/or other possible genetic risks on a volunteer basis.¹⁸ The American Medical Association agrees with that position and states that "*genetic testing must be voluntary, informed consent must be required, and confidentiality must be maintained*".¹⁹ The NIH Cancer Genetics Studies Consortium further suggests that medical follow-up and counseling is recommended if a genetic mutation is found after genetic testing.²⁰ Genetic testing for breast cancer can lead to interventions that result in the diagnosis of early stages of breast cancer.²¹

Great concern has been expressed about the commercialization of pre-symptomatic genetic testing, by organizations such as the American Society of Human Genetics, The National Advisory Council of Human Genome Research and the National Action Plan for Breast Cancer. Results of these tests can be harmful because there is currently no known way to avoid cancer and no cure. Women who test negative for mutated breast cancer genes may falsely feel they are at no risk for cancer as 90 % of those with breast cancer do not have mutated breast cancer genes²². Research does show that prophylactic mastectomies may reduce the incidence of breast cancer in persons with a family history of breast cancer by 90%, however, this does not preclude the person developing cancer in other parts of the body.²³ Thus, predictive knowledge could do more harm than good.²⁴

Citations

- ¹ Approved by the Rhode Island Commission on Women on April 14, 2003. Approved by the RICW Health Committee on March 24, 2003.
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- ³ Rhode Island Commission on Women, 2001. Position paper on Genetics. Adopted by the Rhode Island Commission on Women on April 1, 2002.
- ⁴ Rhode Island Commission on Women, 2001. Position paper on Quality of Health Care. Adopted by the Rhode Island Commission on Women on March 12, 2001.
- ⁵ National Breast Cancer Coalition. *Positions, Fact Sheets and Analyses*. National Breast Cancer Coalition, 2002 c. Accessed 29 October 2002. Internet. Available from <http://www.natlbcc.org/bin/index.asp?strid=363&depid=9>
- ⁶ National Breast Cancer Coalition. "Presymptomatic Genetic Testing for Heritable Breast Cancer Risk." *National Cancer Institute* 2002 b Accessed 29 October 2002: 1-2. Internet. Available from <http://www.cancer.gov/cancerinfo/doc.aspx?viewid=3c5effc8-7047-464e-9a44-ce02e92d0de3>.
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- ⁸ Rhode Island General Assembly. *An Act Relating to Insurance – Genetic Testing*. S0803 Substitute A, 2001, 1.
- ⁹ Rhode Island General Assembly. #7149 Substitute as Amended. *An Act Relating to Labor Relations – Genetic Testing as a Condition of Employment*, 2002. Accessed 10 Feb 2003. www.rilin.state.ri.us/statutes/statutes.html.
- ¹⁰ National Human Genome Research Institute. *Ethical, Legal, and Social Implications (ELSI) Program Education* Accessed October 29, 2002. Internet. Available from http://www.nhgri.nih.gov/About_NHGRI/Der/ELSI/elsi_ed.html.
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- ¹² US Department of Energy, 1998. *To Know Ourselves*. Office of Health and Environmental Research, 1998. Accessed 30 July 1998. Internet. Available from http://www.lbl.gov/Publications/TKO/08_ethical.htm.
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- ¹⁷ National Academy of Sciences & Institute of Medicine, 1994. Assessing Genetic Risks: Implications for Health Policy from

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- ¹⁸ World Health Organization. Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services. Geneva: World Health Organization, 1997.
- ¹⁹ American Medical Association. *American Medical Association: Policy Compendium*. Chicago: American Medical Association, 1997, 78-9.
- ²⁰ Burke, Wylie, Mary Daly, Judy Garber, Jeffrey Botkin, and Mary Jo Kahn. "Recommendations for Follow-up Care of Individuals with an Inherited Predisposition to Cancer." *Journal of the American Medical Association*, 277 no. 12 (1997): 997-1003.
- ²¹ Dr. Kenneth Offit, Chief of Clinical Genetics at Memorial Sloane-Kettering. Cited in Women's Health Weekly, 2002. Women with BRCA mutations benefited from counseling, screening and surgery. Found in COPYRIGHT 2002, NewsRX, article # A84262186.
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